

What is **WAS?**

Wiskott Aldrich syndrome

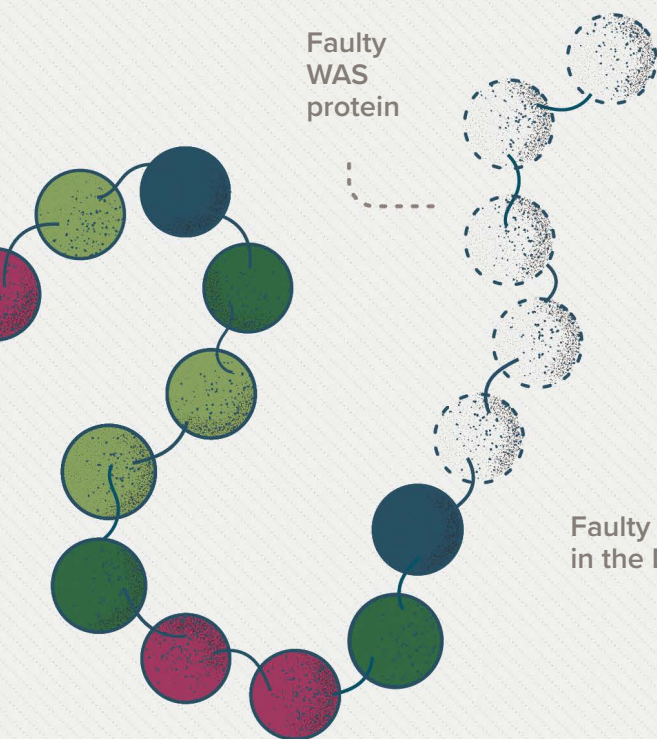
A rare and
life-threatening
inherited immune
disorder



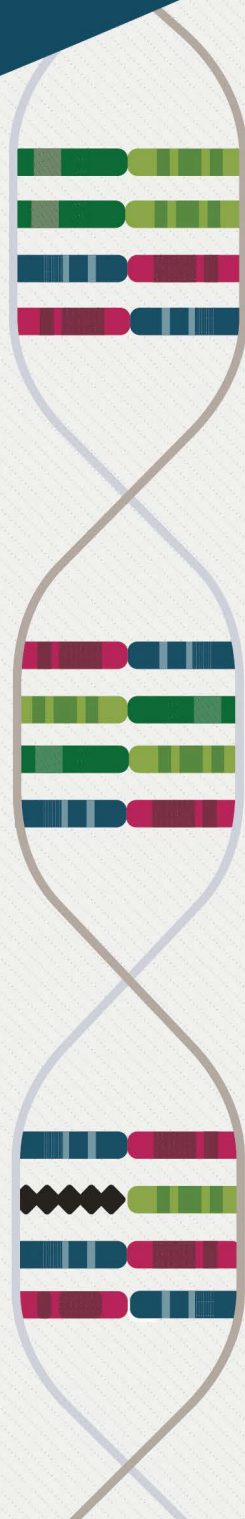
What are the causes of WAS?

WAS is caused by a mutation in the gene that produces the WAS protein, which results in abnormal function of white blood cells, the body's cells responsible for fighting infections.

In addition, WAS leads to reduced size and number of platelets, the cells that are responsible for helping the body to form blood clots to stop bleeding.



Faulty gene in the DNA



How is WAS inherited?

Approximately
**1-4 in every
1,000,000**

**baby boys are
born with WAS**

X-linked recessive inheritance

WAS is inherited in an X-linked pattern, which means that WAS usually only affects boys.

Carriers of the mutation have sometimes been shown to have mild symptoms, similar to those of WAS.



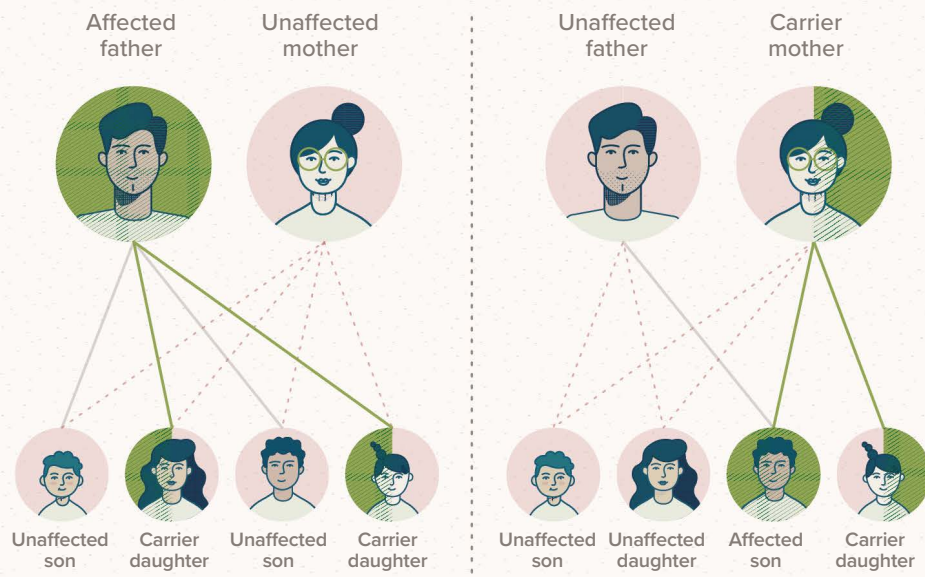
Affected



Unaffected



Carrier



How does WAS affect the body?

Boys with WAS are at risk of serious bleeding and life-threatening infections.

WAS may also lead to an increased risk of several autoimmune- mediated diseases in which the body attacks itself, inflammatory disorders and some cancers (including lymphoma and leukemia).

For more information about WAS and its management, please consult your healthcare provider.



Symptoms may include:

Repeated infections

Serious bleeding

Severe Eczema

Bloody diarrhea

Easy bruising

Tiny red spot rashes called petechiae

Useful terms

Genes

Small sections of DNA that contain the instructions for individual characteristics, like eye and hair color, and how to make proteins, the functional building blocks of the cell. Proteins are responsible for making sure that the cells in the body function properly.

Autoimmune disease

Caused when the immune system attacks healthy cells in the body.

Eczema

A skin rash which can be red, itchy, scaly and distressing.

Immune disorder

A dysfunction of the immune system.

Immune system

Defends the body against foreign invaders, such as bacteria, viruses and fungi.

Microthrombocytopenia

A reduction in the number and size of platelets.

Petechiae (pi-TEE-kee-ee)

Tiny red spots caused by bleeding under the skin.

Infection

The invasion and multiplication of micro-organisms, such as bacteria, viruses and fungi, which may cause symptoms or harmful effects within the body.

Platelets (thrombocytes)

Colorless blood cells that help blood to clot. Platelets stop bleeding by clumping and forming plugs in blood vessel injuries.

Mutation

A change in the structure of a gene or group of genes. Such changes can be passed on from parent to child. Many mutations cause no harm but others can cause genetic disorders, such as WAS.

White blood cells (lymphocytes/leukocytes)

A type of blood cell that plays an important role in the immune system's response to infection. White blood cells are formed in the bone marrow.

X-linked recessive inheritance

A form of inheritance where the mutation, or disease, usually affects only boys.

Thrombocytopenia

A condition in which a person has a low blood platelet count.

References

- Buchbinder D et al. Appl Clin Genet 2014;7:55–66
- Genetics Home Reference [Internet]. Bethesda (MD): The Library Published: June 23, 2020. Available from <https://ghr.nlm.nih.gov/primer/inheritance/inheritancepatterns>

